



PLCB4 gene

phospholipase C beta 4

Normal Function

The *PLCB4* gene provides instructions for making one form (the beta 4 isoform) of a protein called phospholipase C. This protein is involved in a signaling pathway within cells known as the phosphoinositide cycle, which helps transmit information from outside the cell to inside the cell. Phospholipase C carries out one particular step in the phosphoinositide cycle: the conversion of a molecule called phosphatidylinositol 4,5-bisphosphate (PIP_2) to two smaller molecules, inositol 1,4,5-trisphosphate (IP_3) and 1,2-diacylglycerol. These smaller molecules relay messages into the cell that ultimately influence many cell activities.

Studies suggest that the beta 4 isoform of phospholipase C contributes to the development of the first and second pharyngeal arches. These embryonic structures ultimately develop into the jawbones, facial muscles, middle ear bones, ear canals, outer ears, and related tissues. This protein is also thought to play a role in vision, particularly in the function of the retina, which is a specialized tissue at the back of the eye that detects light and color.

Health Conditions Related to Genetic Changes

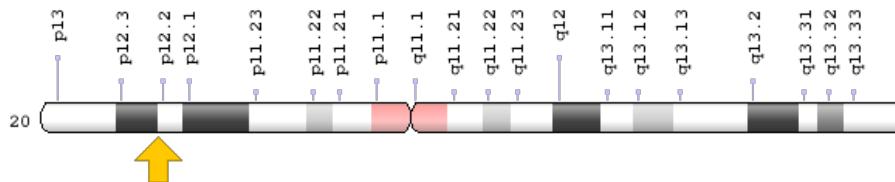
auriculo-condylar syndrome

At least nine mutations in the *PLCB4* gene have been found to cause auriculo-condylar syndrome, a disorder that primarily affects the development of the ears and lower jaw (mandible). The identified mutations change single protein building blocks (amino acids) in the beta 4 isoform of phospholipase C. These changes likely alter the structure of the protein and impair the phosphoinositide cycle. Abnormal signaling alters the formation of the lower jaw: instead of developing normally, the lower jaw becomes shaped more like the smaller upper jaw (maxilla). The abnormal shape leads to an unusually small chin (micrognathia) and problems with jaw function. Researchers are working to determine how mutations in this gene lead to the other developmental abnormalities associated with auriculo-condylar syndrome.

Chromosomal Location

Cytogenetic Location: 20p12.3-p12.2, which is the short (p) arm of chromosome 20 between positions 12.3 and 12.2

Molecular Location: base pairs 9,068,710 to 9,480,816 on chromosome 20 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 1-phosphatidyl-D-myo-inositol-4,5-bisphosphate
- 1-phosphatidylinositol 4,5-bisphosphate phosphodiesterase beta-4
- ARCND2
- dJ1119D9.2 (Phopholipase C, beta 4 (1-Phosphatidylinositol-4,5-Bisphosphate Phosphodiesterase Beta 4))
- inositoltrisphosphohydrolase
- monophosphatidylinositol phosphodiesterase
- phosphoinositidase C
- phosphoinositide phospholipase C-beta-4
- phospholipase C, beta 4
- PI-PLC
- PLC-beta-4
- PLCB4_HUMAN
- triphosphoinositide phosphodiesterase

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): The Hydrolysis of Phosphatidyl Inositol Bisphosphate by Phospholipase C Generates Two Messengers
<https://www.ncbi.nlm.nih.gov/books/NBK22443/>
- Madame Curie Bioscience Database: Phospholipase C
<https://www.ncbi.nlm.nih.gov/books/NBK6267/#A26559>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28PLCB4%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+OR+%28%28phosphoinositidase+C%5BTIAB%5D%29+AND+%28beta+4%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- PHOSPHOLIPASE C, BETA-4
<http://omim.org/entry/600810>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_PLCB4.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=PLCB4%5Bgene%5D>
- HGNC Gene Family: C2 domain containing phospholipases
<http://www.genenames.org/cgi-bin/genefamilies/set/832>
- HGNC Gene Family: Phospholipases
<http://www.genenames.org/cgi-bin/genefamilies/set/467>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9059
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/5332>
- UniProt
<http://www.uniprot.org/uniprot/Q15147>

Sources for This Summary

- Alvarez RA, Ghalayini AJ, Xu P, Hardcastle A, Bhattacharya S, Rao PN, Pettenati MJ, Anderson RE, Baehr W. cDNA sequence and gene locus of the human retinal phosphoinositide-specific phospholipase-C beta 4 (PLCB4). *Genomics*. 1995 Sep 1;29(1):53-61.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/8530101>
- Gordon CT, Vuillot A, Marlin S, Gerkes E, Henderson A, AlKindy A, Holder-Espinasse M, Park SS, Omarjee A, Sanchis-Borja M, Bdira EB, Oufadem M, Sikkema-Raddatz B, Stewart A, Palmer R, McGowan R, Petit F, Delobel B, Speicher MR, Aurora P, Kilner D, Pellerin P, Simon M, Bonnefont JP, Tobias ES, García-Miñaúr S, Bitner-Glindzicz M, Lindholm P, Meijer BA, Abadie V, Denoyelle F, Vazquez MP, Rotky-Fast C, Couloigner V, Pierrot S, Manach Y, Breton S, Hendriks YM, Munnich A, Jakobsen L, Kroisel P, Lin A, Kaban LB, Basel-Vanagaite L, Wilson L, Cunningham ML, Lyonnet S, Amiel J. Heterogeneity of mutational mechanisms and modes of inheritance in auriculocondylar syndrome. *J Med Genet*. 2013 Mar;50(3):174-86. doi: 10.1136/jmedgenet-2012-101331. Epub 2013 Jan 12.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/23315542>
- OMIM: PHOSPHOLIPASE C, BETA-4
<http://omim.org/entry/600810>
- Rieder MJ, Green GE, Park SS, Stamper BD, Gordon CT, Johnson JM, Cunniff CM, Smith JD, Emery SB, Lyonnet S, Amiel J, Holder M, Heggie AA, Bamshad MJ, Nickerson DA, Cox TC, Hing AV, Horst JA, Cunningham ML. A human homeotic transformation resulting from mutations in PLCB4 and GNAI3 causes auriculocondylar syndrome. *Am J Hum Genet*. 2012 May 4;90(5):907-14. doi: 10.1016/j.ajhg.2012.04.002. Erratum in: *Am J Hum Genet*. 2012 Jun 8;90(6):1116. *Am J Hum Genet*. 2012 Aug 10;91(2):397.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/22560091>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3376493/>
- Vines CM. Phospholipase C. *Adv Exp Med Biol*. 2012;740:235-54. doi: 10.1007/978-94-007-2888-2_10. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/22453945>

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/PLCB4>

Reviewed: January 2013

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services